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Mental Retardation, Postaxial Polydactyly, Phalangeal Hypoplasia, 2–3 Toe Syndactyly, Unusual Face, Uncombable Hair: New Syndrome?

K. Kozlowski[1*] and M. Krajewska[2]

[1]*Departments of Radiology and Medical Genetics, Royal Alexandra Hospital for Children, Sydney, Australia*
[2]*Monument Hospital, Child Health Centre, Warsaw, Poland*

We report on a boy with unique somatic and skeletal manifestations. The syndrome consists of mental retardation, postaxial polydactyly, phalangeal hypoplasia, 2–3 toe syndactyly, abnormal face and uncombable hair. A younger sib who died soon after the birth was probably also affected. Am. J. Med. Genet. 68:142–146, 1997 © 1997 Wiley-Liss, Inc.

KEY WORDS: mental retardation; polydactyly; syndactyly; phalangeal hypoplasia; uncombable hair

INTRODUCTION

The boy described is the first child of healthy, nonconsanguineous, young Polish parents. He was born at 38 weeks of gestation by caesarian section. The pregnancy was complicated by vaginal bleeding at 8 months. Birth length was not recorded. Birth weight was 2,500 g. He remained in an intensive care unit for several days. His subsequent psychomotor development was delayed. A sixth finger was removed postaxially in the first year of life.

He was sent to the Child Health Center at the age of 20 months for malformations of unknown cause (Figs. 1A, 2A). His weight was 6.7 kg (<3rd centile), length 68 cm (<3rd centile), OFC 44.5 cm (<3rd centile). The child was developmentally delayed and had bossing, hypotelorism, narrow palpebral fissures with slightly mongoloid slant, prominent nasal root, narrow nasal bridge, narrow lips, micrognathia, abnormal ears with prominent antihelix, poorly folded helix, underdeveloped lobule and antitragus, high palate, short hyperlordotic neck with decreased mobility, pectus carinatum, dysplastic widely spaced nipples, inguinal hernia, dysplastic scrotum, cryptorchism, small bowed penis. The hands were small with short fingers and clinodactyly of the fifth finger. Single palmar crease was present at the left hand. There was equino-valgus deformity of the feet with syndactyly of toes 2 and 3. The big toe was large and broad. His hair were soft, thin and the hair growth was poor.

He was reviewed at ages 2, 10 and 15 years (Figs. 1B–E, 2B–F). His weight and height were <3rd centile. IQ was 60 at the age of 6 and 70 at the age of 10. He had conductive deafness. At the age of 10 years orchiopexy was performed. The testicles were described as small and hard. Hair growth became normal but the hair became uncombable. The eyebrows were thin. There was progressive thoracic kyphosis. He complained of recurrent middle ear and pulmonary infections. Cardiologic and ophthalmologic findings were normal.

At the age of 15 years his weight was 43.4 kg, height 140.2 cm, OFC 52.5 cm. Appearance had changed; his face became elongated, the forehead narrow and prognatism of the lower jaw appeared. His hair became normal on touch but uncombable. Electron microscopic examination (10 hairs) showed longitudinal grooves in 3 hairs and deformities of the tunic in two 2 hairs; 7 were normal (Fig. 3). Blood and urine were normal. Normal karyotype XY. The radiographic examinations performed between 20 months and 15 years showed short phalanges, the middle phalanges being most severely affected. In the chest there was fusion of the segments of the corpus sterni. The ribs were thin, curved inferiorly in the posterior part and oblique in position. The sternal end of the clavicles were widened. The proximal femoral epiphyses were large, the knee epiphyses slightly flattened and the distal tibial epiphyses wedge shaped.

Since then, the parents had another boy who died in a rural hospital at the age of 4 months with the diagnosis of chondrodystrophy and multiple malformations. The mother claims that the infant was similar to his brother.

DISCUSSION

The malformations as presented in our patient do not correspond to any of the postaxial polydactyly syndromes

*Correspondence to: Dr. K. Kozlowski, New Children's Hospital, Westmead, NSW 2145, Australia.

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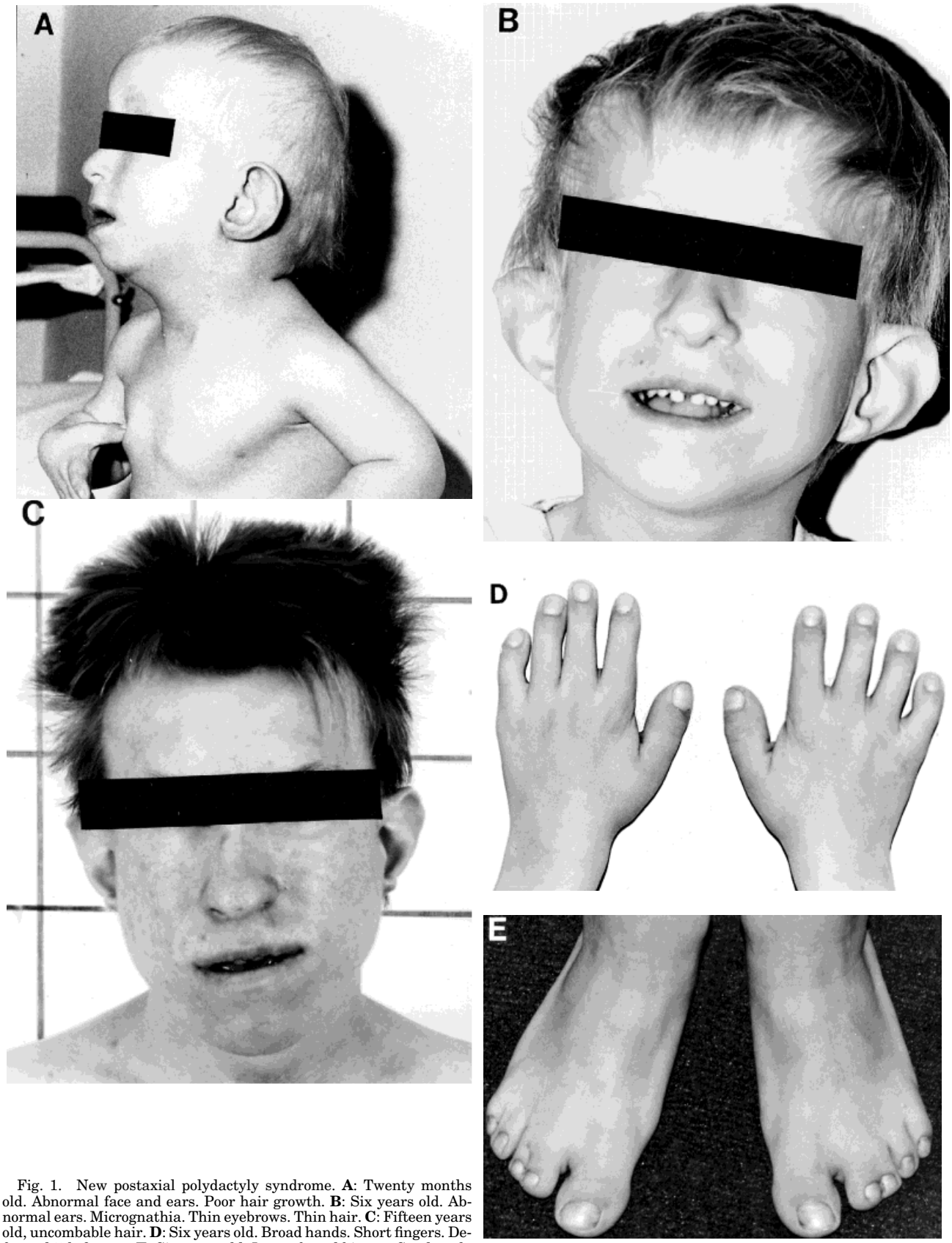

Fig. 1. New postaxial polydactyly syndrome. **A**: Twenty months old. Abnormal face and ears. Poor hair growth. **B**: Six years old. Abnormal ears. Micrognathia. Thin eyebrows. Thin hair. **C**: Fifteen years old, uncombable hair. **D**: Six years old. Broad hands. Short fingers. Deformed 5th fingers. **E**: Six years old. Large, broad big toe. Syndactyly between 2nd and 3rd toes.

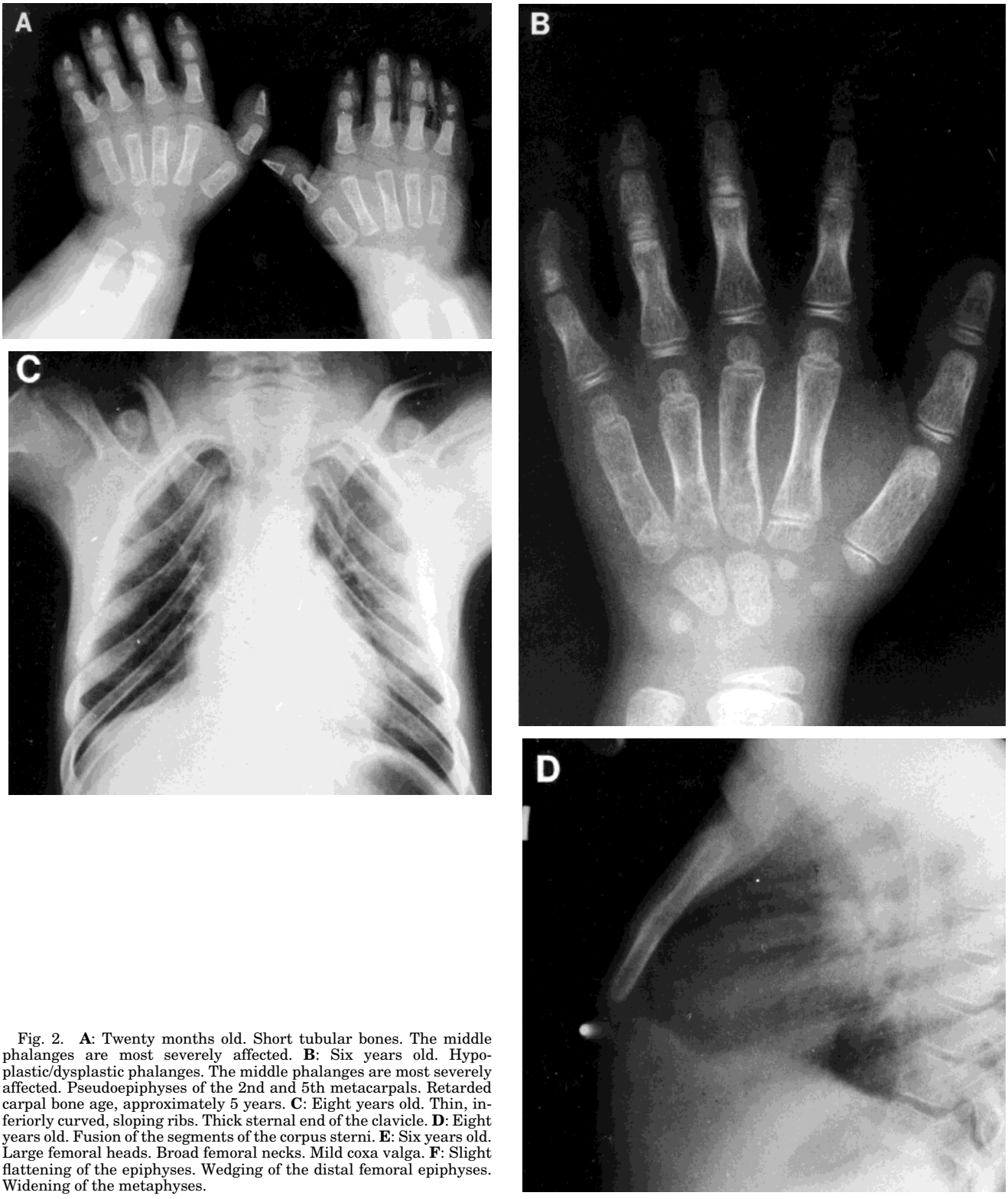

Fig. 2. **A**: Twenty months old. Short tubular bones. The middle phalanges are most severely affected. **B**: Six years old. Hypoplastic/dysplastic phalanges. The middle phalanges are most severely affected. Pseudoepiphyses of the 2nd and 5th metacarpals. Retarded carpal bone age, approximately 5 years. **C**: Eight years old. Thin, inferiorly curved, sloping ribs. Thick sternal end of the clavicle. **D**: Eight years old. Fusion of the segments of the corpus sterni. **E**: Six years old. Large femoral heads. Broad femoral necks. Mild coxa valga. **F**: Slight flattening of the epiphyses. Wedging of the distal femoral epiphyses. Widening of the metaphyses.

Fig. 2. (*continued*).

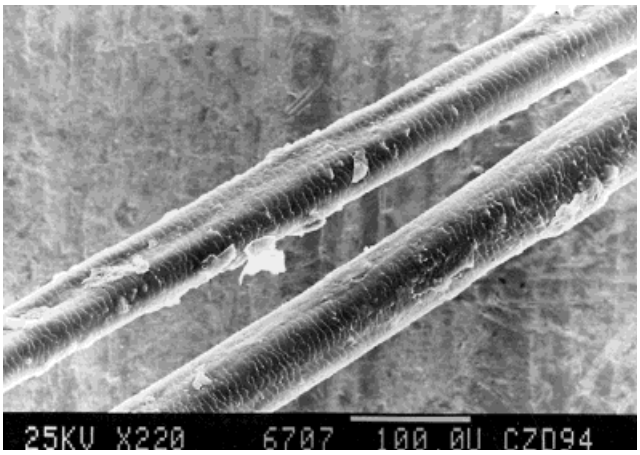

Fig. 3. Scanning electron microscopy shows longitudinal groove in the hair shaft and deformities of the tunic.

[Buyse, 1990; Gorlin et al., 1990]. There is some similarity to the "new postaxial polydactyly syndrome" of Temtamy and McKusick [1978]; however, their patient showed fusion of lower medial and lateral incisors, cleft palate, hemivertebrae with agenesis of 11 and 12 ribs and congenital heart defects anomalies absent in our patient. The family described by Goldberg and Pashayan [1976] "hallux syndactyly—ulnar polydactyly—abnormal ear lobules: A new syndrome" showed syndactyly of the 1st and 2nd toes. The ear malformation was different, a deep horizontal grove. There were different grades of postaxial polydactyly in the family. There was a separate delta phalanx located at the medial aspect of the 1st MTP joint and a hypoplastic metatarsal located between the normal 1st and 2nd metatarsal. Beside postaxial polydactyly these abnormalities were absent in our patient. None of the former patients had abnormal hair. We think that our patient

presents a hitherto unreported malformation syndrome. As the sib was probably also affected, recessive, autosomal or X-linked inheritance is most likely.

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